Bruno Dallapiccola

List of Publications by Year
in descending order

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564 papers	36,084 citations	4641 85 h-index	5227 165 g-index
572 all docs	572 docs citations	572 times ranked	39092 citing authors

#	Article	IF	Citations
1	Hereditary Early-Onset Parkinson's Disease Caused by Mutations in PINK1. Science, 2004, 304, 1158-1160.	6.0	3,060
2	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. Cell, 2001, 107, 513-523.	13.5	2,055
3	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. Nature Genetics, 2007, 39, 1007-1012.	9.4	624
4	Gut microbiota profiling of pediatric nonalcoholic fatty liver disease and obese patients unveiled by an integrated metaâ€omicsâ€based approach. Hepatology, 2017, 65, 451-464.	3.6	572
5	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. Nature Genetics, 2007, 39, 75-79.	9.4	523
6	Mandibuloacral Dysplasia Is Caused by a Mutation in LMNA-Encoding Lamin A/C. American Journal of Human Genetics, 2002, 71, 426-431.	2.6	509
7	Common variants at five new loci associated with early-onset inflammatory bowel disease. Nature Genetics, 2009, 41, 1335-1340.	9.4	459
8	PINK1 mutations are associated with sporadic early-onset parkinsonism. Annals of Neurology, 2004, 56, 336-341.	2.8	447
9	Transmembrane 6 superfamily member 2 gene variant disentangles nonalcoholic steatohepatitis from cardiovascular disease. Hepatology, 2015, 61, 506-514.	3.6	424
10	Mitochondrial import and enzymatic activity of PINK1 mutants associated to recessive parkinsonism. Human Molecular Genetics, 2005, 14, 3477-3492.	1.4	413
11	Development and Validation of a Multidimensional Prognostic Index for One-Year Mortality from Comprehensive Geriatric Assessment in Hospitalized Older Patients. Rejuvenation Research, 2008, 11, 151-161.	0.9	397
12	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidyl inositol signaling to the ciliopathies. Nature Genetics, 2009, 41, 1032-1036.	9.4	383
13	Parkes Weber syndrome, vein of Galen aneurysmal malformation, and other fast-flow vascular anomalies are caused byRASA1 mutations. Human Mutation, 2008, 29, 959-965.	1.1	382
14	Grouping of Multiple-Lentigines/LEOPARD and Noonan Syndromes on the PTPN11 Gene. American Journal of Human Genetics, 2002, 71, 389-394.	2.6	380
15	Mutations in the Pericentrin (PCNT) Gene Cause Primordial Dwarfism. Science, 2008, 319, 816-819.	6.0	370
16	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature Genetics, 2006, 38, 623-625.	9.4	368
17	Cystinuria caused by mutations in rBAT, a gene involved in the transport of cystine. Nature Genetics, 1994, 6, 420-425.	9.4	366
18	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. Nature Genetics, 2009, 41, 1022-1026.	9.4	358

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19	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
20	Joubert Syndrome and related disorders. Orphanet Journal of Rare Diseases, 2010, 5, 20.	1.2	325
21	The origin of the major cystic fibrosis mutation (Î”F508) in European populations. Nature Genetics, 1994, 7, 169-175.	9.4	323
22	Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrodental dysostosis. Nature Genetics, 2000, 24, 283-286.	9.4	323
23	Deletion of KDM6A, a Histone Demethylase Interacting with MLL2, in Three Patients with Kabuki Syndrome. American Journal of Human Genetics, 2012, 90, 119-124.	2.6	315
24	A restricted spectrum of NRAS mutations causes Noonan syndrome. Nature Genetics, 2010, 42, 27-29.	9.4	271
25	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	9.4	261
26	GermlineBRAFmutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: Molecular diversity and associated phenotypic spectrum. Human Mutation, 2009, 30, 695-702.	1.1	251
27	Leopard syndrome. Orphanet Journal of Rare Diseases, 2008, 3, 13.	1.2	250
28	Further Delineation of Deletion 1p36 Syndrome in 60 Patients: A Recognizable Phenotype and Common Cause of Developmental Delay and Mental Retardation. Pediatrics, 2008, 121, 404-410.	1.0	233
29	Mutations in lectin complement pathway genes COLEC11 and MASP1 cause 3MC syndrome. Nature Genetics, 2011, 43, 197-203.	9.4	229
30	Heterozygous Germline Mutations in the CBL Tumor-Suppressor Gene Cause a Noonan Syndrome-like Phenotype. American Journal of Human Genetics, 2010, 87, 250-257.	2.6	221
31	Congenital heart diseases in children with Noonan syndrome: An expanded cardiac spectrum with high prevalence of atrioventricular canal. Journal of Pediatrics, 1999, 135, 703-706.	0.9	216
32	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	9.4	211
33	Additive Effects of Genetic Variation in Dopamine Regulating Genes on Working Memory Cortical Activity in Human Brain. Journal of Neuroscience, 2006, 26, 3918-3922.	1.7	208
34	Germline Missense Mutations Affecting KRAS Isoform B Are Associated with a Severe Noonan Syndrome Phenotype. American Journal of Human Genetics, 2006, 79, 129-135.	2.6	205
35	Mutations in ANKRD11 Cause KBG Syndrome, Characterized by Intellectual Disability, Skeletal Malformations, and Macrodontia. American Journal of Human Genetics, 2011, 89, 289-294.	2.6	205
36	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	2.6	204

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37	Prenatal Diagnosis of Myotonic Dystrophy Using Fetal DNA Obtained from Maternal Plasma. Clinical Chemistry, 2000, 46, 301-302.	1.5	201
38	The human gut microbiota: a dynamic interplay with the host from birth to senescence settled during childhood. Pediatric Research, 2014, 76, 2-10.	1.1	194
39	Natural Gene-Expression Variation in Down Syndrome Modulates the Outcome of Gene-Dosage Imbalance. American Journal of Human Genetics, 2007, 81, 252-263.	2.6	187
40	Survival Motor-Neuron Gene Transcript Analysis in Muscles from Spinal Muscular-Atrophy Patients. Biochemical and Biophysical Research Communications, 1995, 213, 342-348.	1.0	182
41	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. Nature Genetics, 2015, 47, 661-667.	9.4	177
42	AHI1 is required for photoreceptor outer segment development and is a modifier for retinal degeneration in nephronophthisis. Nature Genetics, 2010, 42, 175-180.	9.4	171
43	Reference ranges of HOMA-IR in normal-weight and obese young Caucasians. Acta Diabetologica, 2016, 53, 251-260.	1.2	166
44	Mediterranean Diet and Health: Food Effects on Gut Microbiota and Disease Control. International Journal of Molecular Sciences, 2014, 15, 11678-11699.	1.8	162
45	Searching for Psoriasis Susceptibility Genes in Italy: Genome Scan and Evidence for a New Locus on Chromosome 1. Journal of Investigative Dermatology, 1999, 112, 32-35.	0.3	161
46	Genetic Susceptibility to Nonsteroidal Anti-Inflammatory Drugâ€“Related Gastroduodenal Bleeding: Role of Cytochrome P450 2C9 Polymorphisms. Gastroenterology, 2007, 133, 465-471.	0.6	161
47	Prevalence and Clinical Significance of Cardiovascular Abnormalities in Patients With the LEOPARD Syndrome. American Journal of Cardiology, 2007, 100, 736-741.	0.7	150
48	Multidimensional Prognostic Index Based on a Comprehensive Geriatric Assessment Predicts Short-Term Mortality in Older Patients With Heart Failure. Circulation: Heart Failure, 2010, 3, 14-20.	1.6	146
49	NF1 Gene Mutations Represent the Major Molecular Event Underlying Neurofibromatosis-Noonan Syndrome. American Journal of Human Genetics, 2005, 77, 1092-1101.	2.6	139
50	Mutant Pink1 induces mitochondrial dysfunction in a neuronal cell model of Parkinsonâ€™s disease by disturbing calcium flux. Journal of Neurochemistry, 2009, 108, 1561-1574.	2.1	139
51	Gut Microbiota Markers in Obese Adolescent and Adult Patients: Age-Dependent Differential Patterns. Frontiers in Microbiology, 2018, 9, 1210.	1.5	139
52	Gerodermia osteodysplastica is caused by mutations in SCYL1BP1, a Rab-6 interacting golgin. Nature Genetics, 2008, 40, 1410-1412.	9.4	138
53	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113.	2.6	137
54	Anatomic patterns of conotruncal defects associated with deletion 22q11. Genetics in Medicine, 2001, 3, 45-48.	1.1	135

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55	The INSL3-LGR8/GREAT Ligand-Receptor Pair in Human Cryptorchidism. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 4273-4279.	1.8	134
56	Guidelines for the appropriate use of genetic tests in infertile couples. European Journal of Human Genetics, 2002, 10, 303-312.	1.4	129
57	LEOPARD syndrome: Clinical diagnosis in the first year of life. American Journal of Medical Genetics, Part A, 2006, 140A, 740-746.	0.7	129
58	Mutations ofZFPM2/FOG2 gene in sporadic cases of tetralogy of Fallot. Human Mutation, 2003, 22, 372-377.	1.1	127
59	Genotypes and phenotypes of Joubert syndrome and related disorders. European Journal of Medical Genetics, 2008, 51, 1-23.	0.7	127
60	Familial transposition of the great arteries caused by multiple mutations in laterality genes. Heart, 2010, 96, 673-677.	1.2	126
61	Assignment of a locus for autosomal dominant idiopathic scoliosis (IS) to human chromosome 17p11. Human Genetics, 2002, 111, 401-404.	1.8	125
62	AHI1gene mutations cause specific forms of Joubert syndrome-related disorders. Annals of Neurology, 2006, 59, 527-534.	2.8	125
63	Correlation between cardiac involvement and CTG trinucleotide repeat length in myotonic dystrophy. Journal of the American College of Cardiology, 1995, 25, 239-245.	1.2	124
64	Diversity, parental germline origin, and phenotypic spectrum of de novoHRASmissense changes in Costello syndrome. Human Mutation, 2007, 28, 265-272.	1.1	123
65	Hennekam syndrome can be caused by FAT4 mutations and be allelic to Van Maldergem syndrome. Human Genetics, 2014, 133, 1161-1167.	1.8	122
66	The search for South European cystic fibrosis mutations: Identification of two new mutations, four variants, and intronic sequences. Genomics, 1991, 10, 193-200.	1.3	117
67	22q11 deletions in isolated and syndromic patients with tetralogy of Fallot. Human Genetics, 1995, 95, 479-82.	1.8	117
68	Gut Microbiota Dysbiosis as Risk and Premorbid Factors of IBD and IBS Along the Childhoodâ€“Adulthood Transition. Inflammatory Bowel Diseases, 2016, 22, 487-504.	0.9	117
69	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. Human Molecular Genetics, 2014, 23, 4315-4327.	1.4	114
70	Complete Transposition of the Great Arteries. Circulation, 2001, 104, 2809-2814.	1.6	113
71	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. European Journal of Human Genetics, 2015, 23, 1068-1071.	1.4	113
72	Cayler cardiofacial syndrome and del22qll: Part of the CATCH22 phenotype. American Journal of Medical Genetics Part A, 1994, 53, 303-304.	2.4	110

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73	A Functional Variant of the Adipocyte Glycerol Channel Aquaporin 7 Gene Is Associated With Obesity and Related Metabolic Abnormalities. Diabetes, 2007, 56, 1468-1474.	0.3	108
74	Germline and somatic NF1 mutations in sporadic and NF1â€associated malignant peripheral nerve sheath tumours. Journal of Pathology, 2009, 217, 693-701.	2.1	107
75	Gut microbiota signatures in cystic fibrosis: Loss of host CFTR function drives the microbiota enterophenotype. PLoS ONE, 2018, 13, e0208171.	1.1	107
76	Congenital heart defects in Kabuki syndrome. American Journal of Medical Genetics Part A, 2001, 100, 269-274.	2.4	105
77	Recurrent microdeletion at 17q12 as a cause of Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome: two case reports. Orphanet Journal of Rare Diseases, 2009, 4, 25.	1.2	105
78	Distinguishing the four genetic causes of jouberts syndrome-related disorders. Annals of Neurology, 2005, 57, 513-519.	2.8	104
79	The Functional Q84R Polymorphism of Mammalian Tribbles Homolog TRB3 Is Associated With Insulin Resistance and Related Cardiovascular Risk in Caucasians From Italy. Diabetes, 2005, 54, 2807-2811.	0.3	100
80	Park6-linked parkinsonism occurs in several european families. Annals of Neurology, 2002, 51, 14-18.	2.8	98
81	Mutations in PVRL4, Encoding Cell Adhesion Molecule Nectin-4, Cause Ectodermal Dysplasia-Syndactyly Syndrome. American Journal of Human Genetics, 2010, 87, 265-273.	2.6	98
82	A homozygousGJA1 gene mutation causes a Hallermann-Streiff/ODDD spectrum phenotype. Human Mutation, 2004, 23, 286-286.	1.1	97
83	SOS1 mutations in Noonan syndrome: molecular spectrum, structural insights on pathogenic effects, and genotype-phenotype correlations. Human Mutation, 2011, 32, 760-772.	1.1	97
84	MKS3/TMEM67mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. Human Mutation, 2009, 30, E432-E442.	1.1	96
85	Keppen-Lubinsky Syndrome Is Caused by Mutations in the Inwardly Rectifying K+ Channel Encoded by KCNJ6. American Journal of Human Genetics, 2015, 96, 295-300.	2.6	95
86	Description, Nomenclature, and Mapping of a Novel Cerebello-Renal Syndrome with the Molar Tooth Malformation. American Journal of Human Genetics, 2003, 73, 663-670.	2.6	91
87	Copy-Number Variations Involving the IHH Locus Are Associated with Syndactyly and Craniosynostosis. American Journal of Human Genetics, 2011, 88, 70-75.	2.6	89
88	Gut Microbiota Profiling and Gutâ€“Brain Crosstalk in Children Affected by Pediatric Acute-Onset Neuropsychiatric Syndrome and Pediatric Autoimmune Neuropsychiatric Disorders Associated With Streptococcal Infections. Frontiers in Microbiology, 2018, 9, 675.	1.5	88
89	Genetically Determined Interaction between the Dopamine Transporter and the D₂Receptor on Prefronto-Striatal Activity and Volume in Humans. Journal of Neuroscience, 2009, 29, 1224-1234.	1.7	87
90	UFD1L, a Developmentally Expressed Ubiquitination Gene, is Deleted in CATCH 22 Syndrome. Human Molecular Genetics, 1997, 6, 259-265.	1.4	85

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91	BRF1 mutations alter RNA polymerase IIIâ€“dependent transcription and cause neurodevelopmental anomalies. Genome Research, 2015, 25, 155-166.	2.4	85
92	POPDC1S201F causes muscular dystrophy and arrhythmia by affecting protein trafficking. Journal of Clinical Investigation, 2015, 126, 239-253.	3.9	85
93	Phylogenetic and Metabolic Tracking of Gut Microbiota during Perinatal Development. PLoS ONE, 2015, 10, e0137347.	1.1	84
94	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adamsâ€“Oliver Syndrome With Variable Cardiac Anomalies. Circulation: Cardiovascular Genetics, 2015, 8, 572-581.	5.1	84
95	Deletion of a 5-cM Region at Chromosome 8p23 Is Associated With a Spectrum of Congenital Heart Defects. Circulation, 2000, 102, 432-437.	1.6	83
96	Atypical deletions suggest five 22q11.2 critical regions related to the DiGeorge/velo-cardio-facial syndrome. European Journal of Human Genetics, 1999, 7, 903-909.	1.4	82
97	An ATG Repeat in the 3â€²-Untranslated Region of the Human Resistin Gene Is Associated with a Decreased Risk of Insulin Resistance. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 4403-4406.	1.8	82
98	Type 2 Deiodinase Polymorphism (Threonine 92 Alanine) Predicts l-Thyroxine Dose to Achieve Target Thyrotropin Levels in Thyroidectomized Patients. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 910-913.	1.8	82
99	Deficiency for the Ubiquitin Ligase UBE3B in a Blepharophimosis-Ptosis-Intellectual-Disability Syndrome. American Journal of Human Genetics, 2012, 91, 998-1010.	2.6	82
100	Expression of Î”F508 CFTR in normal mouse lung after site-specific modification of CFTR sequences by SFHR. Gene Therapy, 2001, 8, 961-965.	2.3	81
101	Comparison of occurrence of genetic syndromes in ventricular septal defect with pulmonic stenosis (classic tetralogy of Fallot) versus ventricular septal defect with pulmonic atresia. American Journal of Cardiology, 1996, 77, 1375-1376.	0.7	80
102	Fine Mapping of the PSORS4 Psoriasis Susceptibility Region on Chromosome 1q21. Journal of Investigative Dermatology, 2001, 116, 728-730.	0.3	80
103	Cardiac malformations in patients with oral-facial-skeletal syndromes: Clinical similarities with heterotaxia. , 1999, 84, 350-356.		79
104	Mutation spectrum of MLL2 in a cohort of kabuki syndrome patients. Orphanet Journal of Rare Diseases, 2011, 6, 38.	1.2	79
105	The Multidimensional Prognostic Index Predicts Short- and Long-Term Mortality in Hospitalized Geriatric Patients With Pneumonia. Journals of Gerontology - Series A Biological Sciences and Medical Sciences, 2009, 64A, 880-887.	1.7	78
106	Genetics of Pediatric Obesity. Pediatrics, 2012, 130, 123-133.	1.0	78
107	Incidence of chromosome abnormalities and clinical significance of karyotype in de novo acute myeloid leukemia. Cancer Genetics and Cytogenetics, 1993, 67, 28-34.	1.0	77
108	Mutations of the Nogo-66 receptor (RTN4R) gene in schizophrenia. Human Mutation, 2004, 24, 534-535.	1.1	77

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109	The Multidimensional Prognostic Index (MPI), Based on a Comprehensive Geriatric Assessment Predicts Short- and Long-Term Mortality in Hospitalized Older Patients with Dementia. Journal of Alzheimer's Disease, 2009, 18, 191-199.	1.2	77
110	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	1.1	77
111	A Restricted Spectrum of Mutations in the SMAD4 Tumor-Suppressor Gene Underlies Myhre Syndrome. American Journal of Human Genetics, 2012, 90, 161-169.	2.6	77
112	Low doses of dexamethasone constantly delivered by autologous erythrocytes slow the progression of lung disease in cystic fibrosis patients. Blood Cells, Molecules, and Diseases, 2004, 33, 57-63.	0.6	76
113	Familial recurrence of congenital heart disease: an overview and review of the literature. European Journal of Pediatrics, 2006, 166, 111-116.	1.3	76
114	Epistasis between Dopamine Regulating Genes Identifies a Nonlinear Response of the Human Hippocampus During Memory Tasks. Biological Psychiatry, 2008, 64, 226-234.	0.7	76
115	Benchmarks for Cystic Fibrosis carrier screening: A European consensus document. Journal of Cystic Fibrosis, 2010, 9, 165-178.	0.3	75
116	Joubert syndrome and related disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1879-1888.	1.0	75
117	Cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. International Journal of Cardiology, 2017, 245, 92-98.	0.8	75
118	PINK1heterozygous rare variants: prevalence, significance and phenotypic spectrum. Human Mutation, 2008, 29, 565-565.	1.1	74
119	Spectrum of MEK1 and MEK2 gene mutations in cardio-facio-cutaneous syndrome and genotypeâ€“phenotype correlations. European Journal of Human Genetics, 2009, 17, 733-740.	1.4	74
120	Prostaglandin transporter mutations cause pachydermoperiostosis with myelofibrosis. Human Mutation, 2012, 33, 1175-1181.	1.1	74
121	A 4â€Polymorphism Risk Score Predicts Steatohepatitis in Children With Nonalcoholic Fatty Liver Disease. Journal of Pediatric Gastroenterology and Nutrition, 2014, 58, 632-636.	0.9	74
122	Associated cardiac anomalies in isolated and syndromic patients with tetralogy of fallot. American Journal of Cardiology, 1996, 77, 505-508.	0.7	73
123	Changes in CpG Islands Promoter Methylation Patterns during Ductal Breast Carcinoma Progression. Cancer Epidemiology Biomarkers and Prevention, 2009, 18, 2694-2700.	1.1	73
124	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 621-630.	2.6	73
125	Erythrocytes-Mediated Delivery of Dexamethasone in Steroid-Dependent IBD Patients-A Pilot Uncontrolled Study. American Journal of Gastroenterology, 2005, 100, 1370-1375.	0.2	71
126	Interstitial 22q13 deletions: genes other than SHANK3 have major effects on cognitive and language development. European Journal of Human Genetics, 2008, 16, 1301-1310.	1.4	71

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127	Sex Differences in the Association of Apolipoprotein E and Angiotensin-Converting Enzyme Gene Polymorphisms With Healthy Aging and Longevity: A Population-Based Study From Southern Italy. Journals of Gerontology - Series A Biological Sciences and Medical Sciences, 2006, 61, 918-923.	1.7	70
128	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. Human Genetics, 1992, 89, 653-658.	1.8	69
129	Kabuki syndrome: clinical and molecular diagnosis in the first year of life. Archives of Disease in Childhood, 2015, 100, 158-164.	1.0	69
130	Sequence-specific modification of genomic DNA by small DNA fragments. Journal of Clinical Investigation, 2003, 112, 637-641.	3.9	68
131	SHOX duplications found in some cases with type I Mayer-Rokitansky-Kuster-Hauser syndrome. Genetics in Medicine, 2010, 12, 634-640.	1.1	67
132	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. Human Mutation, 2015, 36, 1080-1087.	1.1	67
133	Atrioventricular canal defect without Down syndrome: A heterogeneous malformation. American Journal of Medical Genetics Part A, 1999, 85, 140-146.	2.4	66
134	Whole gene deletion and splicing mutations expand thePINK1 genotypic spectrum. Human Mutation, 2007, 28, 98-98.	1.1	66
135	Erythrocyte-Mediated Delivery of Dexamethasone in Patients With Mild-to-Moderate Ulcerative Colitis, Refractory to Mesalamine: A Randomized, Controlled Study. American Journal of Gastroenterology, 2008, 103, 2509-2516.	0.2	66
136	Biallelic Mutations in TBCD , Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 962-973.	2.6	66
137	Evidence for Interaction between Psoriasis-Susceptibility Loci on Chromosomes 6p21 and 1q21. American Journal of Human Genetics, 1999, 65, 1798-1800.	2.6	64
138	Novel and recurrent EVC and EVC2 mutations in Ellis-van Creveld syndrome and Weyers acrofacial dyostosis. European Journal of Medical Genetics, 2013, 56, 80-87.	0.7	64
139	Common fragile sites: Their prevalence in subjects with constitutional and acquired chromosomal instability. American Journal of Medical Genetics Part A, 1987, 27, 471-482.	2.4	63
140	Copy-Number Variation of the Glucose Transporter Gene SLC2A3 and Congenital Heart Defects in the 22q11.2 Deletion Syndrome. American Journal of Human Genetics, 2015, 96, 753-764.	2.6	62
141	Parental origin of chromosome 4p deletion in Wolf-Hirschhorn syndrome. American Journal of Medical Genetics Part A, 1993, 47, 921-924.	2.4	61
142	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. American Journal of Human Genetics, 2015, 97, 99-110.	2.6	61
143	Targeted Correction of a Defective Selectable Marker Gene in Human Epithelial Cells by Small DNA Fragments. Molecular Therapy, 2001, 3, 178-185.	3.7	60
144	Congenital heart defects in molecularly proven Kabuki syndrome patients. American Journal of Medical Genetics, Part A, 2017, 173, 2912-2922.	0.7	60

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145	Deletion 8p syndrome. , 1998, 75, 534-536.		59
146	Spinal muscular atrophy genotyping by gene dosage using multiple ligation-dependent probe amplification. Neurogenetics, 2006, 7, 269-276.	0.7	59
147	Diagnosis of Noonan syndrome and related disorders using target next generation sequencing. BMC Medical Genetics, 2014, 15, 14.	2.1	59
148	Nonrandom association of atrioventricular canal and del (8p) syndrome. American Journal of Medical Genetics Part A, 1992, 42, 424-427.	2.4	58
149	Analysis of the ?-sarcoglycan gene in familial and sporadic myoclonus-dystonia: Evidence for genetic heterogeneity. Movement Disorders, 2003, 18, 1047-1051.	2.2	58
150	TheTRIB3Q84R Polymorphism and Risk of Early-Onset Type 2 Diabetes. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 190-196.	1.8	58
151	cDNA characterization and chromosomal mapping of two human homologues of the Drosophila dishevelled polarity gene. Human Molecular Genetics, 1996, 5, 953-958.	1.4	57
152	Epilepsy with auditory features: ALGI1 gene mutation suggests a loss-of-function mechanism. Annals of Neurology, 2003, 53, 396-399.	2.8	57
153	Genotype and cardiovascular phenotype correlations with TBX1 in 1,022 velo-cardio-facial/digeorge/22q11.2 deletion syndrome patients. Human Mutation, 2011, 32, 1278-1289.	1.1	57
154	An excess of chromosome 1 breakpoints in male infertility. European Journal of Human Genetics, 2004, 12, 993-1000.	1.4	56
155	Three patients with oculoâ€auriculoâ€vertebral spectrum and microdeletion 22q11.2. American Journal of Medical Genetics, Part A, 2009, 149A, 2860-2864.	0.7	56
156	The Pittâ€Hopkins syndrome: Report of 16 new patients and clinical diagnostic criteria. American Journal of Medical Genetics, Part A, 2011, 155, 1536-1545.	0.7	55
157	Transposition of the great arteries associated with deletion of chromosome 22q11. American Journal of Cardiology, 1995, 75, 95-98.	0.7	54
158	Orphanet Journal of Rare Diseases: Launch Editorial. Orphanet Journal of Rare Diseases, 2006, 1, 1.	1.2	54
159	Assessing the role ofDRD5 andDYT1 in two different caseâ€“control series with primary blepharospasm. Movement Disorders, 2007, 22, 162-166.	2.2	54
160	High-resolution SNP arrays in mental retardation diagnostics: how much do we gain?. European Journal of Human Genetics, 2010, 18, 178-185.	1.4	54
161	The internet user profile of Italian families of patients with rare diseases: a web survey. Orphanet Journal of Rare Diseases, 2013, 8, 76.	1.2	54
162	Deletion 22q11 in patients with interrupted aortic arch. American Journal of Cardiology, 1999, 84, 360-361.	0.7	53

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163	Analysis of Clinically Relevant Single-Nucleotide Polymorphisms by Use of Microelectronic Array Technology. Clinical Chemistry, 2002, 48, 2124-2130.	1.5	53
164	Arrayâ€based comparative genomic hybridization in earlyâ€stage mycosis fungoides: Recurrent deletion of tumor suppressor genes BCL7A, SMAC/DIABLO, and RHOF. Genes Chromosomes and Cancer, 2008, 47, 1067-1075.	1.5	53
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